

<120> MUTATIONS IN AND GENOMIC STRUCTURE OF HERG - A LONG QT
SYNDROME GENE

<130> 2323-136

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<150> 09/122,847

<151> 1998-07-27

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<170> PatentIn Ver. 2.0

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Pro Gly Ala Val Val Val Asp Val Asp Leu Thr Pro Ala Ala Pro Ser
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Ser Glu Ser Leu Ala Leu Asp Glu Val Thr Ala Met Asp Asn His Val
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Ala Gly Leu Gly Pro Ala Glu Glu Arg Arg Ala Leu Val Gly Pro Gly
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tct ccg ccc cgc agc gcg ccc ggc cag ctc cca tcg ccc cgg gcg cac 828
Ser Pro Pro Arg Ser Ala Pro Gly Gln Leu Pro Ser Pro Arg Ala His
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agc ctc aac ccc gac gcc tcg ggc tcc agc tgc agc ctg gcc cgg acg 876
 Ser Leu Asn Pro Asp Ala Ser Gly Ser Ser Cys Ser Leu Ala Arg Thr
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cgc tcc cga gaa agc tgc gcc agc gtg cgc cgc gcc tcg tcg gcc gac 924
 Arg Ser Arg Glu Ser Cys Ala Ser Val Arg Arg Ala Ser Ser Ala Asp
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gac atc gag gcc atg cgc gcc ggg gtg ctg ccc ccg cca ccg cgc cac 972
 Asp Ile Glu Ala Met Arg Ala Gly Val Leu Pro Pro Pro Pro Arg His
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gcc agc acc ggg gcc atg cac cca ctg cgc agc ggc ttg ctc aac tcc 1020
 Ala Ser Thr Gly Ala Met His Pro Leu Arg Ser Gly Leu Leu Asn Ser
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 Thr Ser Asp Ser Asp Leu Val Arg Tyr Arg Thr Ile Ser Lys Ile Pro
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caa atc acc ctc aac ttt gtg gac ctc aag ggc gac ccc ttc ttg gct 1116
 Gln Ile Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala
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 Ser Pro Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg
 355 360 365

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 Thr His Asn Val Thr Glu Lys Val Thr Gln Val Leu Ser Leu Gly Ala
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 Asp Val Leu Pro Glu Tyr Lys Leu Gln Ala Pro Arg Ile His Arg Trp
 385 390 395

acc atc ctg cat tac agc ccc ttc aag gcc gtg tgg gac tgg ctc atc 1308
 Thr Ile Leu His Tyr Ser Pro Phe Lys Ala Val Trp Asp Trp Leu Ile
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 415 420 425 430

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 Phe Leu Leu Lys Glu Thr Glu Glu Gly Pro Pro Ala Thr Glu Cys Gly
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 Tyr Ala Cys Gln Pro Leu Ala Val Val Asp Leu Ile Val Asp Ile Met
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 Phe Ile Val Asp Ile Leu Ile Asn Phe Arg Thr Thr Tyr Val Asn Ala
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aac gag gag gtg gtc agc cac ccc ggc cgc atc gcc gtc cac tac ttc 1548
 Asn Glu Glu Val Val Ser His Pro Gly Arg Ile Ala Val His Tyr Phe
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Lys Gly Trp Phe Leu Ile Asp Met Val Ala Ala Ile Pro Phe Asp Leu
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Leu Ile Phe Gly Ser Gly Ser Glu Glu Leu Ile Gly Leu Leu Lys Thr
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Ala Arg Leu Leu Arg Leu Val Arg Val Ala Arg Lys Leu Asp Arg Tyr
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Ser Glu Tyr Gly Ala Ala Val Leu Phe Leu Leu Met Cys Thr Phe Ala
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Leu Ile Ala His Trp Leu Ala Cys Ile Trp Tyr Ala Ile Gly Asn Met
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Asp Gln Ile Gly Lys Pro Tyr Asn Ser Ser Gly Leu Gly Gly Pro Ser
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Ile Phe Gly Asn Val Ser Ala Ile Ile Gln Arg Leu Tyr Ser Gly Thr
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gcc cgc tac cac aca cag atg ctg cgg gtg cgg gag ttc atc cgc ttc 2124
Ala Arg Tyr His Thr Gln Met Leu Arg Val Arg Glu Phe Ile Arg Phe
675 680 685

cac cag atc ccc aat ccc ctg cgc cag cgc ctc gag gag tac ttc cag 2172
His Gln Ile Pro Asn Pro Leu Arg Gln Arg Leu Glu Glu Tyr Phe Gln
690 695 700

cac gcc tgg tcc tac acc aac gcc atc gac atg aac gcg gtg ctg aag 2220
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705 710 715

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00735995-434400

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Ser	Arg	Gly	Ser	Ile	Glu	Ile	Leu	Arg	Gly	Asp	Val	Val	Val	Ala	Ile	
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Pro	Gly	Lys	Ser	Asn	Gly	Asp	Val	Arg	Ala	Leu	Thr	Tyr	Cys	Asp	Leu	
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His	Lys	Ile	His	Arg	Asp	Asp	Leu	Leu	Glu	Val	Leu	Asp	Met	Tyr	Pro	
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Thr	Asp	Lys	Asp	Thr	Glu	Gln	Pro	Gly	Glu	Val	Ser	Ala	Leu	Gly	Pro	
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Trp	Gly	Glu	Ser	Pro	Ser	Ser	Gly	Pro	Ser	Ser	Pro	Glu	Ser	Ser	Glu	
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Glu	Leu	Pro	Pro	Gly	Ala	Pro	Glu	Leu	Pro	Gln	Glu	Gly	Pro	Thr	Arg		
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Arg	Leu	Ser	Leu	Pro	Gly	Gln	Leu	Gly	Ala	Leu	Thr	Ser	Gln	Pro	Leu		
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His	Arg	His	Gly	Ser	Asp	Pro	Gly	Ser									
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 Cys Glu Leu Cys Gly Tyr Ser Arg Ala Glu Val Met Gln Arg Pro Cys
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 Thr Cys Asp Phe Leu His Gly Pro Arg Thr Gln Arg Arg Ala Ala Ala
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 Gln Ile Ala Gln Ala Leu Leu Gly Ala Glu Glu Arg Lys Val Glu Ile
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 Ala Phe Tyr Arg Lys Asp Gly Ser Cys Phe Leu Cys Leu Val Asp Val
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 Val Pro Val Lys Asn Glu Asp Gly Ala Val Ile Met Phe Ile Leu Asn
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 Phe Glu Val Val Met Glu Lys Asp Met Val Gly Ser Pro Ala His Asp
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 Thr Asn His Arg Gly Pro Pro Thr Ser Trp Leu Ala Pro Gly Arg Ala
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 Lys Thr Phe Arg Leu Lys Leu Pro Ala Leu Leu Ala Leu Thr Ala Arg
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 Glu Ser Ser Val Arg Ser Gly Gly Ala Gly Gly Ala Gly Ala Pro Gly
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 Ala Val Val Val Asp Val Asp Leu Thr Pro Ala Ala Pro Ser Ser Glu
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 Ser Leu Ala Leu Asp Glu Val Thr Ala Met Asp Asn His Val Ala Gly
 210 215 220
 Leu Gly Pro Ala Glu Glu Arg Arg Ala Leu Val Gly Pro Gly Ser Pro
 225 230 235 240
 Pro Arg Ser Ala Pro Gly Gln Leu Pro Ser Pro Arg Ala His Ser Leu
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 Asn Pro Asp Ala Ser Gly Ser Ser Cys Ser Leu Ala Arg Thr Arg Ser
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 Arg Glu Ser Cys Ala Ser Val Arg Arg Ala Ser Ser Ala Asp Asp Ile
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 Glu Ala Met Arg Ala Gly Val Leu Pro Pro Pro Pro Arg His Ala Ser
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Thr Gly Ala Met His Pro Leu Arg Ser Gly Leu Leu Asn Ser Thr Ser
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 Asp Ser Asp Leu Val Arg Tyr Arg Thr Ile Ser Lys Ile Pro Gln Ile
 325 330 335
 Thr Leu Asn Phe Val Asp Leu Lys Gly Asp Pro Phe Leu Ala Ser Pro
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 Thr Ser Asp Arg Glu Ile Ile Ala Pro Lys Ile Lys Glu Arg Thr His
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 Leu His Tyr Ser Pro Phe Lys Ala Val Trp Asp Trp Leu Ile Leu Leu
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 Glu Val Val Ser His Pro Gly Arg Ile Ala Val His Tyr Phe Lys Gly
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 Asp Lys Tyr Val Thr Ala Leu Tyr Phe Thr Phe Ser Ser Leu Thr Ser
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 Val Gly Phe Gly Asn Val Ser Pro Asn Thr Asn Ser Glu Lys Ile Phe
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Ser Ile Cys Val Met Leu Ile Gly Ser Leu Met Tyr Ala Ser Ile Phe
 645 650 655
 Gly Asn Val Ser Ala Ile Ile Gln Arg Leu Tyr Ser Gly Thr Ala Arg
 660 665 670
 Tyr His Thr Gln Met Leu Arg Val Arg Glu Phe Ile Arg Phe His Gln
 675 680 685
 Ile Pro Asn Pro Leu Arg Gln Arg Leu Glu Glu Tyr Phe Gln His Ala
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 Gly Ser Ile Glu Ile Leu Arg Gly Asp Val Val Val Ala Ile Leu Gly
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 820 825 830
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 Pro Arg Pro Pro Gly Glu Pro Pro Gly Gly Glu Pro Leu Met Glu Asp
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Cys Glu Lys Ser Ser Asp Thr Cys Asn Pro Leu Ser Gly Ala Phe Ser
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<223> Description of Artificial Sequence:Hypothetical
sequence for the example of calculating homology.

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<212> DNA

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<223> Description of Artificial Sequence:Hypothetical
sequence for example of calculating homology.

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<400> 82
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Leu Ile Ala His Trp Leu
1 5


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Leu Ile Val His Trp Leu
  1               5
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<211> 6
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<400> 104
Leu Ala Ala His Trp Lys
1 5

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<213> Rattus rattus
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<400> 105
Leu Ala Ala His Trp Met
1 5

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<213> Drosophila melanogaster
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1 5

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Ganetzky, 1994.

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1 5

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<211> 7
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<400> 108
Asp Ile Leu Ile Asn Phe Arg
1 5

[illegible]

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<212> PRT
<213> Homo sapiens
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<400> 109
Asp Ile Leu Ile Asp Phe Arg
1 5

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<212> PRT
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Asp Ile Val Leu Asn Phe His
  1                               5
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<220>
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Ganetzky, 1994.

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<400> 111
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<213> Homo sapiens
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<400> 112
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<213> Homo sapiens
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```
<400> 113
Ser Val Gly Phe Ser Asn Val Ser
  1                      5
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<210> 114
<211> 8
<212> PRT
<213> Mus musculus
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```
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Ser Val Gly Phe Gly Asn Ile Ala
      1               5
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[illegible]

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<212> PRT
<213> Drosophila melanogaster
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<400> 115
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  1                      5
```

```
<210> 116
<211> 8
<212> PRT
<213> Drosophila melanogaster
```

```
<400> 116
Thr Val Gly Tyr Gly Asp Met Thr
  1             5
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[illegible]